

THIS IS NOT A TEST REQUEST FORM.
The information below is required to perform EDS VI testing.
Please fill out this form and submit it with the test request form or electronic packing list.

PATIENT HISTORY FOR EHLERS-DANLOS KYPHOSCOLIOTIC FORM (TYPE VI) TESTING

Patient Name _____ **Date of Birth** ____/____/____ **Gender** F M
Physician _____ **Physician Phone** (____) _____ **Practice Specialty** _____
Genetic Counselor _____ **Counselor Phone** (____) _____

Patient's Ethnicity (check all that apply)

- | | | | |
|---|---|--|--------------------------------------|
| <input type="checkbox"/> African American | <input type="checkbox"/> Ashkenazi Jewish | <input type="checkbox"/> Asian | <input type="checkbox"/> Caucasian |
| <input type="checkbox"/> Hispanic | <input type="checkbox"/> Middle Eastern | <input type="checkbox"/> Native American | <input type="checkbox"/> Other _____ |

Does the patient have SYMPTOMS? No Yes Unknown If yes, check all that apply

<input type="checkbox"/> Kyphoscoliosis	<input type="checkbox"/> Thin hyperextensible skin	<input type="checkbox"/> Arterial rupture
<input type="checkbox"/> Hypotonia	<input type="checkbox"/> Atrophic scarring	<input type="checkbox"/> Respiratory compromise
<input type="checkbox"/> Joint hypermobility	<input type="checkbox"/> Other _____	

LABORATORY FINDINGS

Deoxyypyridinoline: Pyridinoline crosslinks in urine: Normal Abnormal Not performed Unknown
 List ratio: _____

Lysyl hydroxylase activity in fibroblasts: Normal Abnormal Not performed Unknown
 List percent activity: _____

Collagen screening in fibroblasts: Normal Abnormal Not performed Unknown

Does the patient have a FAMILY HISTORY OF EDS VI? No Yes

If yes, attach a PEDIGREE or specify the **RELATIONSHIP** of family member(s) to the patient. _____

Is the relative a healthy carrier or affected with EDS VI?

What are the *PLODI* mutations in the family member? _____

Circle the test you intend to order.

0080351 Ehlers-Danlos Syndrome Type VI Screen: Urine screen, ordered as first line test before DNA.

2005559 Ehlers-Danlos Syndrome Kyphoscoliotic Form, Type VI (*PLOD1*) Sequencing and Deletion/Duplication:
 Sequencing and deletion/duplication analysis of *PLOD1* coding regions and intron/exon boundaries. Clinical sensitivity of this test is unknown.

2005555 Ehlers-Danlos Syndrome Kyphoscoliotic Form, Type VI (*PLOD1*) Deletion/Duplication: Deletion/duplication analysis of *PLOD1* coding regions. Clinical sensitivity of this test is approximately 20%.

2001961 Familial Mutation, Targeted Sequencing: A copy of the relative's DNA lab result is REQUIRED for this test.

For questions, contact an ARUP genetic counselor at (800) 242-2787, ext. 2141

Master Label